Infant & Toddler
Connection of Virginia

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To: Local System Managers

From: Mary Ann Discenza

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Subj: Expanded Newborn Screening Services

Effective March 2006, the Virginia Newborn Screening Services program, through the Virginia Department of Health, was expanded to include testing for 28 heritable disorders and genetic diseases. Infants under 6 months of age who are born in Virginia will be screened for the following disorders and diseases, which are identified through newborn dried blood-spot screening tests.

1. Argininosuccinic acidemia (ASA);
2. Beta-Ketothiolase deficiency (ßKT);
3. Biotinidase deficiency (BIOT);
4. Carnitine uptake defect (CUD);
5. Citrullinemia (CIT);
6. Congenital adrenal hyperplasia (CAH);
7. Congenital hypothyroidism (CH);
8. Cystic fibrosis (CF);
9. Galactosemia (GALT);
10. Glutaric acidemia type I (GA I);
11. Hemoglobin Sickle/Beta-thalassemia (Hb S/ßTh);
12. Hemoglobin Sickle/C disease (Hb S/C);
13. Homocystinuria (HCY);
14. Isovaleric acidemia (IVA);
15. Long chain hydroxyacyl-CoA dehydrogenase deficiency (LCHAD);
16. Maple syrup urine disease (MSUD);
17. Medium-chain acyl-CoA dehydrogenase deficiency (MCAD);
18. Methylmalonic acidemia (mutase deficiency) (MUT);
19. Methylmalonic acidemia (Cbl A,B);
20. Multiple carboxylase deficiency (MCD);
21. Phenylketonuria (PKU);
22. Propionic acidemia (PROP);
23. Sickle cell anemia (Hb SS disease) (Hb SS);
24. Tyrosinemia type I (TYR I);
25. Trifunctional protein deficiency (TFP);
26. Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD);

27. 3-hydroxy 3-methyl glutaric aciduria (HMG), and
28. 3-Methylcrotonyl-CoA carboxylase deficiency (3MCC).

Any infant whose parent or guardian objects on the grounds that the tests conflict with his religious practices or tenets will not be required to receive the newborn dried blood-spot screening tests.

**All of the disorders and conditions listed above are considered diagnosed conditions with a high probability of resulting in developmental delay. Therefore, these infants will be eligible under Part C in Virginia.** When entering these infants into ITOTS, please note the following:

- Congenital adrenal hyperplasia and sickle cell anemia should be listed as "other" under Diagnosed Disabling Condition.
- All other disorders and diseases listed above should be listed as inborn errors of metabolism. (Note: The Department of Health has stated that, although congenital hypothyroidism is currently given in the Individual Child Data Form instructions as an example of an inborn error of metabolism, it would be more appropriate to list this condition as an endocrine disorder. This change will be made when enhancements to the ITOTS system are made. Until then, please continue to list it as an inborn error of metabolism in accordance with current instructions.]

A fact sheet about the disorders and diseases added to the newborn screening system in 2006 was developed by Alexandra Iwashyna, a medical intern who worked with the Part C Office this spring, and is attached. You will also find helpful information about the disorders and diseases listed above, including parent guides, at http://www.vahealth.org/genetics/servgp.htm.